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Letter to the Editor

Toriello-Carey Syndrome

To the Editor:

Toriello-Carey syndrome was described in 1988. Czarnecki et al. reviewed the cases published in 1996. They point out an unbalanced sex ratio. We report on another male case reinforcing their hypothesis of an X-linked or sex-influenced gene.

Our patient was born to a 25-year-old primigravid woman at 35 weeks of gestation. Pregnancy was uncomplicated until 30 weeks when intrauterine growth retardation and breech presentation were found. Birth weight was 1,950 g. Apgar scores were 6, 5, and 8 at 1, 5, and 10 minutes, respectively. Because of respiratory failure, reanimation was necessary. At birth he was noted to have microretrognathia, large posterior clef palate and glossoptosis, hypertelorism, short palpebral fissures, small nose, apparently low-set ears, short neck with excess skin, narrow chest, and congenital hypotonia.

Chest roentgenograph showed a left cervical rib. Echocardiography documented a ventricular septal defect (VSD), an atrial septal defect (ASD), and a thick ventricular septum. Cerebral magnetic resonance imaging (MRI) scan showed partial agenesis of corpus callosum and cortical atrophy.

Chromosomes were apparently normal; no 22q11 deletion was found. Muscle biopsy showed nonspecific abnormalities. The patient died at 2 months when life support was stopped. Autopsy was refused.

Toriello-Carey syndrome was suspected because of clinical and radiological findings. There is no consanguinity in this family. A second pregnancy was monitored and cystic hygroma was diagnosed by echography at 12 weeks. Fetal karyotype was normal (46,XY). Spontaneous abortion occurred at 15 weeks. A second occurrence of Toriello-Carey syndrome was suspected but remains unconfirmed.

This case, in addition to the 10 cases reviewed by Czarnecki et al., accentuates the difference between male and female involvement. We support the hypothesis of X-linked inheritance postulated by these authors.

REFERENCES

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